SurePrint Community Design Prenatal CGH+SNP 8X60K

Streamline your prenatal research with a CNV and LOH all-in-one assay

Advantages of the design

- Detection of CNVs and LOH in a single assay
- Streamlined analysis
- Developed by experts in the field
- Regions of interest specific for prenatal research
- Simplified data interpretation

CGH+SNP Microarrays for prenatal research

Chromosomal microarray analysis (CMA) is considered a front-line test for the analysis of genetic anomalies in prenatal samples related to developmental disabilities. It is endorsed by the American College of Medical Genetics and Genomics, the Child Neurology Society, the American Academy of Neurology, and the European Guidelines for cytogenetic analysis[1,2,3,4].

Genetic anomalies can be the major etiology of various constitutional disorders such as cleft lip, congenital birth defects, intellectual disability (ID), and developmental delay (DD). Copy number variants (CNVs) are one of the most common genetic variations. Many CNVs have been found to statistically increase the risk of DD, ID, and have recently been identified in autism spectrum disorder.

CGH+SNP Microarrays have a unique combination of probes targeting genomic regions and single nucleotide polymorphisms (SNPs). This combination of probe types makes it possible to simultaneously detect CNVs and copy-neutral changes (such as uniparental disomy) on the same array.

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The Agilent SurePrint CD Prenatal CGH+SNP 8X60K microarray can be used to detect a wide variety of aberrations in prenatal samples. These include single gene gains and losses, amplifications, and deletions relevant for prenatal clinical research. In addition, to evaluate regions of loss of heterozygosity (LOH), the design includes a subset of the Agilent SNP probe set covering all chromosomes at low resolution.

The focused design of the SurePrint CD Prenatal CGH+SNP array delivers single-gene or single-exon resolution on well-known microdeletion/microduplication syndromes, enabling accurate detection of various genetic aberrations. Combined with Agilent CGH streamlined sample preparation, high-quality results are achievable within two to three days.

The unique design of the SurePrint CD Prenatal CGH+SNP microarray allows researchers to screen high-resolution copy number changes in 152 genomic regions. The design ensures low-resolution LOH coverage in all chromosomes and a genome-wide backbone with a median probe spacing of 78 Kb, all of which are key for prenatal clinical research.

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Table 1. List of syndromic regions selected to be covered by the design.

OMIM Entities	Chromosome Band	Involved Genes
Jacobsen syndrome	11q24-q25	*multiple*
OTODENTAL DYSPLASIA	11q13	*FGF3*
NRXN1 deletion syndrome	2p16	*NRXN1*
15q26 deletion syndrome (IGF1R)	15q26	*IGF1R*
Split-hand/foot malformation 5	2q31	*unknown*
Angelman syndrome	15q11.2-q13	*UBE3A*
Chr 2q37 deletion syndrome	2q37	*multiple*
Holoprosencephaly 6	2q37.1-q37.3	*HPE6*
Chr 10q22.3-q23.2 deletion syndrome	10q22.3-q23.2	*multiple*
Cat eye syndrome	22q11	*unknown*
CHR 15q25 DELETION SYNDROME	15q25	–
Beckwith-Wiedemann syndrome	11p15.4-p15.5	*ICR1, KCNQ1OT1, CDKN1C*
DiGeorge syndrome/velocardiofacial syndrome complex-2	10p14-p13	*unknown*
CHR 17p13.3, TELOMERIC, DUPLICATION SYNDROME	17p13.3-p13.1	–
Chr 16p12.2-p11.2 deletion syndrome	16p11.2-p12.1	*multiple*
Trichorhinophalangeal syndrome, type II	8q24.11-q24.13	*multiple (TRPS1, EXT1)*
MENTAL RETARDATION, AUTOSOMAL DOMINANT 20; MRD20	5q14.3	*MEF2C*
5q14.3 deletion syndrome (MEF2C)	5q14.3	*MEF2C*
Chr 2p16.1-p15 deletion syndrome	2p15-p16.1	*multiple*
Split-hand/foot malformation 3	10q24	*multiple*
Alpha-thalassemia/mental retardation syndrome, type 1	Xq21.1	*ATRX*
Menkes disease	Xq21.1	*ATP7A*
Duchenne muscular dystrophy	Xp21.2-p21.1	*DMD*
Synpolydactyly 1	2q31.1	*HOXD13*
CHR 2q31.1 DUPLICATION SYNDROME	2q31.1	–
Mental retardation, X-linked syndromic, Lubs type	Xq28	MECP2
Mucopolysaccharidosis II	Xq28	IDS
Rett syndrome	Xq28	MECP2
Chr 22q11.2 duplication syndrome	22q11.2	*multiple*
CHR 22q11.2 DELETION SYNDROME, DISTAL	22q11.2	–
22q11.2 distal deletion (BCR, MAPK1)	22q11.2	*BCR; MAPK1*
Rubinstein-Taybi syndrome 1	16p13.3	*CREBBP*
MESOMELIA-SYNOSTOSES SYNDROME	8q13	–
CHR 16p13.3 DUPLICATION SYNDROME	16p13.3	*CREBBP*
Lissencephaly, X-linked Subcortical laminal heterotopia, X-linked	Xq23	*DCX*
Lymphoproliferative syndrome, X-linked, 1	Xq25	*SH2D1A*
CHR 5q12 DELETION SYNDROME	5q12	–
46,XX SEX REVERSAL 2; SRXX2	17q24.3-q25.1	*SOX9*
SPINOCEREBELLAR ATAXIA 20; SCA20	11q12	–
Lesch-Nyhan syndrome	Xq26.2-q26.3	*HPRT1*
Linear skin defects with multiple congenital anomalies 1	Xp22.2	*HCCS*
Opitz GBBB syndrome, type I	Xp22.2	*MID1*
Orofaciodigital syndrome I	Xp22.2	*OFD1*
BRAIN MALFORMATIONS WITH OR WITHOUT URINARY TRACT DEFECTS; BRMUTD	1p31.3	*NFIA*
FRIAS SYNDROME	14q22.1-q22.3	*BMP4*
Phelan-McDermid syndrome	22q13.3	*multiple (SHANK3)*
22q13.3 duplication syndrome (SHANK3/PROSAP2)	22q13.3	*SHANK3*
MICROCORIA, CONGENITAL	13q32	–
8p23.1 deletion	8p23.1	*GATA4*
CHR 15q14 DELETION SYNDROME	15q14	MEIS2
Axenfeld-Rieger syndrome, type 1	4q25	PITX2
CHR 17q12 DUPLICATION SYNDROME	17q12	–
CHR 17q12 DELETION SYNDROME	17q12	–
RCAD sydrome (renal cysts and diabetes) (HNF1B or ACACA)	17q12	*HNF1B; ACACA*
Potocki-Lupski syndrome	17p11.2	*multiple*
Smith-Magenis syndrome	17p11.2	*multiple (RAI1)*
Holoprosencephaly 2	2p21	*SIX3*
CHR 17q11.2 DELETION SYNDROME, 1.4-MB	17q11.2	*NF1*
GLASS SYNDROME; GLASS	2q33.1	*SATB2*
Chr 3q29 deletion syndrome	3q29	*multiple*
Chr 3q29 duplication syndrome	3q29	*unknown*
Cleidocranial dysplasia, forme fruste, dental anomalies, with brachydactyly	6p21.1	*RUNX2*
NABLUS MASK-LIKE FACIAL SYNDROME; NMLFS	8q22.1	–
Witteveen-Kolk syndrome	15q24	*SIN3A*
Trichorhinophalangeal syndrome, type I	8q23.3	*TRPS1*
Pitt-Hopkins syndrome (TCF4)	18q21.2	*TCF4*
1q44 deletion syndrome (AKT3)	1q44	*AKT3*
DOWN SYNDROME	21q22.3	*multiple*
Holoprosencephaly 1	21q22.3	*unknown*
sacral/anorectal malformations	6q25.3	*unknown*
Aniridia Cataract with late-onset corneal dystrophy	11p13	*PAX6*
Wilms tumor, aniridia, genitourinary anomalies and mental retardation syndrome	11p13	*multiple (PAX6, WT1)*
Wilms tumor	11p13	*WT1*
CHR 11p13 DELETION SYNDROME, DISTAL	11p13	*ELP4 (606985) and PAX6 (607108)*
11p13 duplication/triplication syndrome (PAX6)	11p13	*PAX6*
Williams-Beuren region duplication syndrome, Chromosome 7q11.23 duplication syndrome	7q11.23	*multiple*
Charcot-Marie-Tooth disease, type 1A	17p12	PMP22
Neuropathy, recurrent, with pressure palsies	17p12	PMP22
Potocki-Shaffer syndrome	11p11.2	multiple
Williams-Beuren syndrome	7q11.23	multiple
Hernia, congenital diaphragmatic 1	15q26.1	*unknown*
Split-hand/foot malformation 1	7q21.3	*multiple (DSS1, DLX5, DLX6)*
Alport syndrome 1, X-linked	Xq22.3	*COL4A5*
Fragile X syndrome	Xq27.3	*FMR1*
Prader-Willi syndrome	15q11.2	*multiple (SNRPN, NDN)*
CHR 15q11.2 DELETION SYNDROME	15q11.2	*TUBGCP5, NIPA1, NIPA2, CYFIP1*
Cardiofaciocutaneous syndrome	7q34	*BRAF*
Dyggve-Melchior-Clausen disease	18q21.1	*DYM*
Mowat-Wilson syndrome	2q22.3	*ZEB2*

Table 1. Continued.

OMIM Entities	Chromosome Band	Involved Genes
Cornelia de Lange syndrome 1	5p13.2	*NIPBL*
Mental retardation, autosomal recessive 59	8q21.13	*IMPA1*
Feingold syndrome 1	2p24.3	*MYCN*
Nail-patella syndrome	9q33.3	*LMX1B*
Wolf-Hirschhorn syndrome	4p16.3	*multiple*
Chr 1q21.1 deletion syndrome	1q21.1	*multiple*
Chr 1q21.1 duplication syndrome	1q21.1	*multiple*
Developmental and epileptic encephalopathy 1, early infantile, 1	Xp21.3	*ARX*
Saethre-Chotzen syndrome with or without eyelid anomalies	7p21.1	*TWIST1*
Thrombocytopenia-absent radius syndrome	1q21.1	*RBM8A*
Waardenburg syndrome, type 2A	3p13	*MITF*
Heterotaxy, visceral, 1, X-linked Congenital heart defects, nonsyndromic, 1, X-linked	Xq26.3	*ZIC3*
CHR 8q21.11 DELETION SYNDROME	8q21.11	–
Sotos syndrome 1	5q35.3	NSD1
Agammaglobulinemia, X-linked 1	Xq22.1	BTK
DiGeorge syndrome	22q11.21	TBX1
Atrial septal defect 7, with or without AV conduction defects	5q35.1	NKX2-5
VELOCARDIOFACIAL SYNDROME; VCFS	22q11.21	TBX1
Holoprosencephaly 4	18p11.31	TGIF
Nephronophthisis 1, juvenile	2q13	*NPHP1*
CHR 17p13.1 DELETION SYNDROME	17p13.1	–
2q13 deletion syndrome	2q13	–
Holoprosencephaly 3	7q36.3	*SHH*
Koolen-De Vries syndrome	17q21.31	*multiple (KANSL1)*
Chr 17q21.31 duplication syndrome	17q21.31	*multiple*
Chr Xp11.3 deletion syndrome	Xp11.3	*multiple (RP2)*
KLEEFSTRA SYNDROME 1	9q34.3	*EHMT1*
Campomelic dysplasia, Acampomelic campomelic dysplasia, Campomelic dysplasia with autosomal sex reversal	17q24.3	*SOX9*
CHR 3q13.31 DELETION SYNDROME	3q13.31	*ZBTB20*
Optic nerve hypoplasia and abnormalities of the central nervous system Microphthalmia, syndromic 3	3q26.33	*SOX2*
Waardenburg syndrome, type 1	2q36.1	*PAX3*

OMIM Entities	Chromosome Band	Involved Genes
Waardenburg syndrome, type 1	2q36.1	*PAX3*
Holoprosencephaly 9	2q14.2	*unknown*
CORNEAL DYSTROPHY, POSTERIOR AMORPHOUS; PACD	12q21.33	–
Ichthyosis, X-linked	Xp22.31	*STS*
CHR 17q23.1-q23.2 DUPLICATION SYNDROME	17q23.1-q23.2	–
Branchiootorenal syndrome 1	8q13.3	*EYA1*
Otofaciocervical syndrome	8q13.3	*multiple (EYA1)*
Lissencephaly 1 Subcortical laminar heterotopia	17p13.3	*PAFAH1B1*
Miller-Dieker lissencephaly syndrome	17p13.3	*multiple*
15q13.2-13.3 deletion (CHRNA7)	15q13.2-13.3	–
Androgen insensitivity syndrome	Xq12	*AR*
Cystinosis, nephropathic Cystinosis, atypical nephropathic	17p13.2	*CTNS*
CHR 19q13.11 DELETION SYNDROME, DISTAL	19q13.11	–
Blepharophimosis, epicanthus inversus, and ptosis	3q22.3	FOXL2
Alagille syndrome 1	20p12.2	JAG1
van der Woude syndrome 1	1p32.2	IRF6
BARAITSER-WINTER SYNDROME 1; BRWS1	7p22.1	ACTB
Basal cell nevus syndrome	9q22.32	*PTCH1*
Holoprosencephaly 5	13q32.3	*ZIC2*
Adrenal hypoplasia, congenital	Xp21.2	*NR0B1*
Glycerol kinase deficiency	Xp21.2	*GK*
Developmental and epileptic encephalopathy 2	Xp22.13	*CDKL5*
X-inactivation, familial skewed	Xq13.2	*XIST*
Noonan syndrome 1	12q24.13	*PTPN11*
16p13.11 duplication	16p13.11	–
Familial adenomatous polyposis 1, Gardner syndrome, Brain tumor-polyposis syndrome 2, Adenomatous polyposis coli	5q22.2	*APC*
Microphthalmia, syndromic 6	14q22.2	*BMP4*
MENTAL RETARDATION, AUTOSOMAL DOMINANT 1; MRD1	2q23.1	*MBD5*
Pelizaeus-Merzbacher disease	Xq22.2	*PLP1*
CHARGE syndrome	8q12.2	*CHD7*
DYRK1A deletion syndrome	21q22.13	*DYRK1A*
Holoprosencephaly 7	9q22.32	*PTCH1*
Forebrain defects	3p21.31	*TDGF1*

Description of the Design

The SurePrint CD Prenatal CGH+SNP 8x60K microarray has multiple features that enable the analysis of clinical prenatal research samples. A brief description of the microarray design is listed below.

Array probe composition	
Type	Number of Features
Control	3,886
CGH	43,081
SNP	16,009

Array probe design	
Region Type	Median Probe Spacing
Overall	22.5 kb
Targeted Regions	5.6 kb
Backbone	79 kb
Telomeric Regions	2.7 kb
SNP Probes	All chromosomes

Developed by an expert

This array originated from the Fetal DNA Chip designed by Dr. Richard Choy, the professor and deputy director of the Prenatal Genetic Diagnosis Centre in the Department of Obstetrics and Gynecology at the Chinese University of Hong Kong. This is the largest public obstetrics and gynecology unit and tertiary referral center in Hong Kong. It serves the 1.7 million people living in HK and oversees nearly 7,000 births annually.

Citations

1. Satya-Murti, S. et al. Chromosomal Microarray Analysis for Intellectual Disabilities. American Academy of Neurology **2013**.
2. Michelson, D. J. et al. Evidence Report: Genetic and Metabolic Testing on Children with Global Developmental Delay, Report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology* **2011**, *77*(17), 1629–1635.
3. South, S.T. et al. ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: Revision 2013. *Genet. Med*. **2013**, *15*(11), 901–909.
4. Marisa Silva et al. European guidelines for constitutional cytogenomic analysis European Journal of Human Genetics, *Eur J Hum Genet*, **2019**, *27*(1), 1-16.

Order information

Product Description	Part Number	# Samples/Kit
SurePrint CD Prenatal CGH+SNP 8X60K	G5988C	8
SureTag Complete DNA Labelling kit	5190-4240	50
SureTag Purification Columns kit	5190-3391	50
Cot-1 Human DNA	5190-3393	312
Hybridization Gasket Slide kit - 8 microarrays per slide format; 5 gasket slides/pack	G2534-60014	40
Oligo aCGH/ChIP-on-chip Hybridization kit	5188-5200	200
Oligo aCGH/ChIP-on-chip Wash Buffer kit	5188-5226	320

Go to http://www.agilent.com/genomics to see all available kit configurations
Note: Agilent has not performed verification and validation on these arrays.

Agilent has not performed verification and validation on the Community Design panels.

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